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Preconception Carrier Screening

Modified in part from ACOG: Preconception Carrier Screening
https://www.acog.org/patient-resources/faqs/pregnancy/carrier-screening

What is preconception carrier screening?
Preconception carrier screening is a type of genetic test you can have before pregnancy that can tell whether you carry a gene for certain genetic disorders. It allows you to find out the chances of having a child with a genetic disorder.

What is a carrier?
For some genetic disorders, it takes two genes for a person to have the disorder. A carrier is a person who has only one gene for the disorder. Carriers have no or mild symptoms but can pass on the gene for that disorder to his or her child.

Who should have carrier screening?
All women who are thinking about becoming pregnant are offered carrier screening for cystic fibrosis, thalassemia, hemoglobinopathies, and spinal musclar atrophy. You can have screening for additional disorders as well. There are two approaches to carrier screening for additional disorders: 1) targeted screening and 2) expanded carrier screening.

What is targeted carrier screening?
In targeted carrier screening, you are tested for disorders based on your ethnicity or family history. If you belong to an ethnic group or race that has a high rate of carriers for a specific genetic disorder, carrier screening for these disorders may be recommended. This also is called ethnic-based carrier screening. If you have a family history of a specific disorder, screening for that disorder may be recommended, regardless of your race or ethnicity.

What is expanded carrier screening?
In expanded carrier screening, many disorders are screened for using a single sample. This type of screening is done without regard to race or ethnicity. Companies that offer expanded carrier screening create their own lists of disorders that they test for. This list is called a screening panel. Some panels tests for more than 100 different disorders. Screening panels usually focus on severe disorders that affect a person's quality of life from an early age.

Is one approach better than the other?
As of 2017, the American College of Obstetricians and Gynecologists (ACOG) recommends either approach is acceptable. But for individuals with a specific family history or ethnicity for certain genetic disorder, a targeted carrier screening would be more appropriate.

Do I have to have carrier screening?
Carrier screening is a voluntary decision. You can choose to have carrier screening or not. There are no right or wrong choices.

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 parnter 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 preconception carrier screening test for all genetic disorders? What carrier screening tests are available?
Carrier screening tests do not detect all genetic disorder. 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 2017, the American College of Obstetricians and Gynecologists (ACOG) recommends carrier screening for cystic fibrosis, spinal muscular atrophy, thalassemia and hemoglobinopathies be offered to all women who are considering pregnancy or are already pregnant 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-postivie 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 crrier 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, followed by preimplantation genetic diagnosis on each of the embryos for the genetic disorder before implanting the embryo into the uterus to achieve a pregnancy. You may also use donor sperm or donor egg to achieve pregnancy. You may choose not to become pregnant.

Who should I speak to if I have more questions about preconcpeption carrier screening?
If you have questions about preconception 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
Carrier Screening for Genetic Conditions
https://www.acog.org/-/media/project/acog/acogorg/clinical/files/committee-opinion/articles/2017/03/carrier-screening-for-genetic-conditions.pdf

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Preconception Carrier Screening

Family Fertility Center offers preconception carrier screening to **all** women of reproductive age. Currently we utilize the following genetic laboratories for preconception carrier screening:

Myriad's Foresight® Carrier Screen screens for 175 genetic disorders, including cystic fibrosis, fragile X, and spinal muscular atrophy. Myriad is in-network with most insurance carriers. 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 with the average out-of-pocket cost is $75 to $160. Myriad 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://myriadwomenshealth.com/patient/myriad-access-program/

Progenity's Preparent Standard or Global Panel Screen tests 29 to 200+ hereditary genetic disorders respectively 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. The exact discount is to be determined by Progenity, and will be based on my personal financial situation. The average patient responsibility was ~$185 in 2017 after deductibles were met. If the female partner is tested positive for one or more of the genetic disorders, Progenity offers testing of the male partner free of charge. https://www.progenity.com/tests/preparent

Natera's Horizon Carrier Screen tests up to 274 + hereditary genetic disorders. Natera is in-network with most insurance carriers. Most patients pay less than $200, many pay zero out of pocket. Natera will run a personalized estimate for in-network patients, those who owe more than $200 will receive a call and will be offered a pay discount. https://www.natera.com/in-network-plans Out of network patients whose coverage is denied in full will pay no more than $200. Natera also offers pre-implantation genetic testing on embryos for $99 if both partners are tested positive carrier for the same genetic disorder. Call or email Wally Zebi: wzebi@natera.com (973)816-3720 for additional questions.

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PLEASE SIGN BELOW TO INDICATE WHETHER YOU WISH TO DECLINE OR PROCEED WITH PRECONCEPTION CARRIER TESTING:

I have been advised and offered to undergo preconception carrier screening. The benefits and risks have been explained and understood by me. I voluntarily decide to **DECLINE.**	**Signature** **Date**
I agree to **PROCEED** with preconception carrier screening with a genetic laboratory of my choice as indicated below.	**Signature** **Date**

Please select one genetic laboratory from the list below to perform your carrier screening test, and initial next to your choice.

		Initial next to the genetic laboratory of your choice.
Myriad*	I understand I shall have 48 hours to decline testing after I have been advised of any costs.	
Progenity*	I understand Progenity will bill my insurance company even though it is out of network. I can request Progenity for a reduced price if my insurance does not cover the test. The exact discount is to be determined by Progenity and will be based on my personal financial situation. The average patient responsibility was ~$185 in 2017 after deductibles were met.	
Natera*	I understand Natera will bill my insurance company even though it is out of network; and that my cost can be reduced to ~$200 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 undergo your preconception carrier screening at any laboratory of your choice. The estimated cost is current as of August of 2018. **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. You are responsible for the cost of any or all of the laboratory or radiologic testing not covered by your insurance.** Family Fertility Center reserves the right to change the testing laboratory without further notice.

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