

## **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 people who are thinking about becoming pregnant are offered carrier screening for cystic fibrosis, thalassemia, hemoglobinopathies, and spinal muscular 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 of 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 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 people 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-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 reproductive 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 preconception 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>

**FAMILY FERTILITY CENTER**

H. Christina Lee, M.D., J.D., H.C.L.D., F.A.C.O.G.

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**Preconception Carrier Screening****FOR THE PATIENT**

Family Fertility Center recommends and offers preconception carrier screening to all people of reproductive age planning to have a child with his/her gamete (sperm or egg). Currently we utilize Natera® ([www.natera.com](http://www.natera.com)) for preconception carrier screening.

**Natera® Horizon Carrier Screen** tests up to 274 hereditary genetic disorders. <https://www.natera.com/womens-health/horizon-advanced-carrier-screening> Natera® is in-network with many insurance carriers including Capital Blue Cross, Blue Shield (PPO), St. Luke's Hospital, Cigna, Keystone Central, and some Geisinger and Aetna plans. Natera® will run a personalized estimate and contact you regarding your out-of-pocket expense. The average out of pocket expense is around \$250 in 2024. Further information about insurance coverage for Natera Horizon Carrier Screen is available at <https://www.natera.com/womens-health/pricing-billing> Natera® offers discount for qualified individuals with its compassionate care program. Check online at [https://patientportal.natera.com/pricing/about\\_compassionate\\_care](https://patientportal.natera.com/pricing/about_compassionate_care) and see if you qualify. See <https://natera.showpad.com/share/M9LjD8QRUNiqabRam8vnU> for more details.

If you decide to proceed with preconception carrier screening with Natera®, we will place an online order with Natera® for you. Natera® will send you an email, a text or a phone call regarding your carrier screening test. **You must respond in order to make arrangement to collect a blood sample from you.** Go to [https://my.natera.com/services/blood\\_draw](https://my.natera.com/services/blood_draw) to find a lab near you.

The turnaround time is about 14 to 21 days. Natera® will notify you by email, text or phone call when your result is available. You can check on the patient portal at <https://my.natera.com/> for your test results. Natera® will also send a copy of your carrier screen results to us, the ordering physician. If the test is positive, your partner must be tested for carrier screen as well. Natera® offers one free session of genetic counseling with a board-certified genetic counselor regardless of your carrier screen results. Natera® also offers pre-implantation genetic testing on embryos at a discounted price if both partners are tested positive carrier for the same genetic disorder.

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

I have been advised and offered to undergo pre-conception carrier screening. The benefits, risks, limitations and cost of carrier screening have been explained and understood by me. I voluntarily decide to <b>DECLINE</b> any preconception carrier screening.	<div style="text-align: right;"> <b>Signature</b>                      <b>Date</b> </div>
I have been advised and offered to undergo pre-conception carrier screening. The benefits, risks, limitations and cost of carrier screening have been explained and understood by me. I understand I have the right to undergo my preconception carrier screening at any laboratory of my choice. I agree to <b>PROCEED</b> with preconception carrier screening with *Natera®.	<div style="text-align: right;"> <b>Signature</b>                      <b>Date</b> </div>

\* You are free to choose any genetic laboratory other than Natera® to perform your carrier screening. Family Fertility Center has no financial relationship with, and does not receive any kick back from any company including Natera®. The estimated cost is current as of Jan 10, 2024. **Family Fertility makes no guarantee the cited cost is up-to-date. It is your responsibility to contact Natera® 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 testing not covered by your insurance.** Family Fertility Center reserves the right to change the testing laboratory without further notice.

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If you decide to proceed with preconception carrier screening with Natera®, we will place an online order with Natera® for you. Natera® will send you an email, a text or a phone call regarding your carrier screening test. **You must respond in order to make arrangement to collect a blood sample from you.** Go to [https://my.natera.com/services/blood\\_draw](https://my.natera.com/services/blood_draw) to find a lab near you.

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