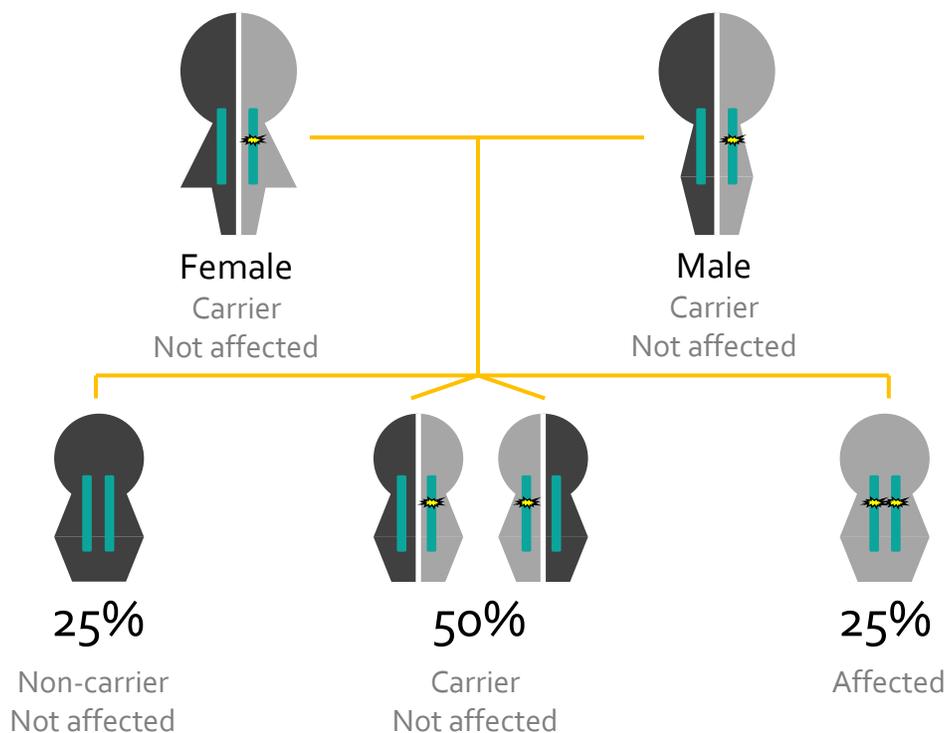


EXPANDED CARRIER SCREENING

We offer expanded carrier screening to patients who are considering pregnancy or who are currently pregnant. Expanded carrier screening is a blood test which looks to see if someone is a “carrier” for a genetic condition. We typically have two copies of each of our genes. A person who is a carrier has a change in one copy of a gene which makes it not work, but that person is not affected because he or she still has the other working copy of the gene. If both members of a couple are carriers for the same genetic condition, they have a 1 in 4 (25%) chance that they will both pass on the non-working gene in each pregnancy they have together. When that happens, that child has no working copies of the gene, and therefore is affected with that condition. People who are carriers usually have no family history, so most people do not know they are carriers until they have a child born with the condition.



The expanded carrier screening test currently offered by PAMF looks for approximately 175 genetic conditions, including Cystic Fibrosis, Tay-Sachs disease, common types of Thalassemia, Spinal Muscular Atrophy, and Fragile X syndrome. Some genetic conditions are very severe and can significantly impair an affected individual’s normal development or even lead to death in childhood. Some require lifelong management. Other conditions are milder and may be treatable. For some conditions, treatment during pregnancy may be available to improve pregnancy outcome.

Like any carrier screening test, some carriers will not be detected. This test also does not look for every possible genetic condition. Therefore this test can reduce, but not eliminate, the chance for a genetic condition.

EXPANDED CARRIER SCREENING

Frequently Asked Questions

Where can I see a list of the genetic disorders screened by expanded carrier screening?

PAMF orders expanded carrier screening through a company called Counsyl. The full list of genetic disorders including descriptions of the conditions is available at www.counsyl.com.

What type of sample is needed for this testing?

Typically expanded carrier screening is done on a single blood draw, however it is also possible to do this testing on saliva.

When is the test performed?

The test can be performed at any time. To have the biggest benefit to you and your partner, it is recommended to have the test done before pregnancy or very early in a pregnancy.

How common is it to be identified as a carrier?

Approximately 60% of individuals who undergo the current expanded carrier screening panel will be identified as a carrier for at least one condition. If one person is identified as a carrier, additional testing will likely be recommended for the other partner. Approximately 2.4% of couples who undergo the current expanded carrier screening panel will be identified as carriers for the same condition, and will thus have a 25% chance to have an affected child in each pregnancy together.

Do the chances to be a carrier change with age?

No, your chance to be a carrier for a genetic condition is not age-dependent.

Do the chances to be a carrier depend on ethnic background?

Some genetic conditions are more common in certain ethnic backgrounds, however people of all ethnicities have a chance to be a carrier of a genetic condition.

What if it is identified that my partner and I are both carriers for the same condition?

If it is identified that you both are carriers for the same condition, you have options. If you are not currently pregnant, you may decide to achieve pregnancy through in vitro fertilization (IVF) with pre-implantation genetic diagnosis (PGD), a pre-pregnancy process that significantly reduces the risk that a child will inherit the genetic disease. During pregnancy, you may decide to undergo diagnostic testing for that condition to make informed reproductive decisions. Some individuals consider adoption, egg or sperm donation, or opt to not have children. Even if you would not choose any of these options, you can use the information to prepare for the birth of a child with a genetic condition. If you are both identified as carriers, you will have the opportunity to speak with your physician or a genetic counselor about the options available to you.

What is the accuracy for expanded carrier screening?

There are many factors which determine the accuracy of carrier screening including your ethnic background and how common a condition is. Expanded carrier screening will not be able to detect everyone who is a carrier, however for most of the conditions it will find over 99% of the carriers. After the test is completed, the expanded carrier screen will provide a residual risk that you could still be a carrier even after testing negative (low-risk) on the screen.

Will my insurance cover the cost for expanded carrier screening?

Typically this testing is covered by insurance, however insurance coverage can be variable. The cost for the test may vary for different insurance companies and plans. PAMF orders expanded carrier screening through a company called Counsyl, which is in-network with most insurance providers. Counsyl can perform an insurance check to provide an estimate of your out of pocket cost. You can call Counsyl directly to request this estimate at 888-COUNSYL (888-268-6795).

Who do I talk to if I'm interested in learning more about expanded carrier screening?

Speak with your healthcare provider or request a referral for you to meet with a prenatal genetic counselor to discuss carrier screening in more detail.