

## Consent for Cystic Fibrosis Screening

Cystic fibrosis (CF) is a life-long illness that severely affects breathing and digestion. It is caused by an abnormal gene that makes the body produce thick mucus in the lungs causing persistent infections. In the pancreas, similar thick secretions can lead to problems with food absorption. Men with CF often have infertility due to absence of the vas deferens, the tubes that conduct sperm from the testes to the urethra. Cystic fibrosis does not affect intelligence or appearance. People with CF have an average lifespan of about 30 years.

Cystic fibrosis is a genetic disorder. Genes are inherited in pairs, one from the mother and one from the father. In CF, both genes must have mutations for a person to manifest the disease. If a person has one altered copy of a CF gene, that person is a **carrier for CF**. There is no known health problems associated with being a carrier. However, if a person has two mutated copies of the CF gene, he or she will develop the disease. When two carriers have a child together, there is a 25% chance that the child will have CF. There is a 50% chance that the child will be a carrier like the parents, and there is a 25% chance that the baby will be completely free of the gene—not a carrier and not have the disease.

About one of every 25 white people (or 4%) carries the altered gene. If your family background is not white, your chance of being a carrier is less than 1 in 30. If you have a relative who is a CF carrier, your risk of being a carrier is greater based on your family history than your ethnic background. You could be a carrier of CF even if no one in your family has the disease.

Ethnicity	Carrier risk	Remaining risk given a negative result
Caucasian, Ashkenazi Jewish	1/25	1/800
Hispanic	1/46	1/150
African-American	1/65	1/240
Asian	1/90	Insufficient data

CF testing screens for the 25 most common types of CF gene mutations (out of several hundreds) as recommended by the American College of Obstetrics & Gynecology. If CF testing shows that both of you are carriers, you may want to see a genetic counselor who can provide more information and help you decide whether to test the baby for CF. Prenatal screening can be done around the 11th week of pregnancy by sampling a tiny piece of the placenta using CVS (chorionic villus sampling), or at the 15th week via amniocentesis in which a needle is used to take fluid from around the baby for testing. If either test shows that the baby will develop CF, you can choose to either terminate or continue the pregnancy.

Couples who both carry the CF mutations can now opt to undergo in vitro fertilization (IVF) instead of natural conception in order to have the embryos tested for CF. Through a procedure called Preimplantation Genetic Diagnosis (PGD), a single cell from each embryo can be removed to test for CF mutations on the third day of culture. Healthy embryos can then be selected for transfer to the woman's uterus.

The cost of CF testing per person is approximately \$300-400 and may not be covered by insurance. You may want to check with your insurance company before deciding if you want testing. Furthermore, if you and your partner would never consider having amniocentesis or CVS (to help you decide about terminating the pregnancy or preparing for the birth of a baby with CF), or IVF/PGD (to help select healthy embryos for transfer), you may not want to be tested.

Patient's Signature \_\_\_\_\_ I **do not** wish to be screened for CF. Should I decide to undergo screening in the future I will contact you on a voluntary basis to be tested.

Patient's Signature \_\_\_\_\_ I desire to be tested for CF and agree to be ultimately responsible for the cost.

Witness: \_\_\_\_\_

Date: \_\_\_\_\_