INFORMED CONSENT / DECLINE FOR CYSTIC FIBROSIS (CF) CARRIER TESTING

Patient name:_____ **D.O.B**:_____ **MR#**:_____

Ethnicity: __ Caucasian __ Jewish __ Hispanic __ African American ___ Asian Other:_____

Family History of CF: ___No ___Yes

Previous CF test: ___No ___Yes (If yes, attach copy of laboratory report)

You should be certain that you understand the following points:

1. Your decision to undergo this test is completely voluntary.
2. You have discussed the information on Cystic Fibrosis carrier testing with your physician or genetic counselor. You have read and understood the Patient Information Sheet on "Carrier Testing for Cystic Fibrosis" that is provided to you.
3. Cystic Fibrosis (CF) is an inherited chronic disease that affects 1 in 3,500 newborns in the US. It affects primarily the lungs and digestive system, specifically the pancreas and the liver, causing the production of very thick, sticky mucus that blocks the air passages and decreases the enzymes necessary for proper digestion of food. It frequently leads to recurrent lung infections and failure to gain weight. The disease is caused by a genetic change or mutation in the cystic fibrosis gene, called the *CFTR* gene, that is passed on to the child from parents who each carry a mutation (carriers). It is a condition that affects either males or females and requires that both copies of the *CFTR* gene pair are defective for someone to have the symptoms. Mutations in the *CFTR* gene are often different depending on the ethnic background. Caucasians and Ashkenazi Jews have the highest carrier risk, followed by Hispanics and African Americans, with Asian Americans having the lowest carrier frequency.
4. The purpose of this test is to determine whether you are a carrier of one of the 39 most common mutations in the *CFTR* gene, which includes the panel of 23 disease-causing mutations recommended by the American College of Medical Genetics-American College of Obstetricians and Gynecologists (see test panel attached). This means that not all mutations in the *CFTR* gene are being detected.
5. This test involves examining the genetic material in a blood sample of at least 5 ml or 1 teaspoonful using the FDA-approved Tag-It Cystic Fibrosis mutation detection method by Luminex 200 xMAP. This detects the 39 mutations in the test panel simultaneously while making a lot of copies of each of the specific mutation.
6. If your test is positive, this means that you have one mutation and this confirms that you are a carrier for CF. Rarely two mutations are identified, which may or may not indicate that you have cystic fibrosis Clinical correlation is necessary.
7. If you are a carrier of a mutation (positive test), testing your partner (the biologic father of the present pregnancy) will help you learn more about the chance that your future baby

could have CF. The clinic and the laboratory need accurate information concerning family history, ethnic background and parentage for a more accurate interpretation of test results.

8. If one parent is a carrier and the other is not, this usually means that your baby will not have CF. However the baby could still be a carrier of the mutation or even very rarely will have CF because of mutations that are not included in the test panel. This can be discussed at length with the genetic counselor.
9. If both parents are carriers, the baby has a 25% risk (or 1 in 4 chances) of having CF. Prenatal testing (performed on the baby while still in the womb) can be done to find out whether or not both of the disease-causing mutations are inherited.
10. If your CF test is negative, this means that it is almost certain that you do not carry one of the 39 most common mutations. However there is still a chance that you may carry one of other mutations not included in the test panel which can still be passed on to any children you may have.
11. Genetic counseling should be sought once you receive the results of this test. Further testing or additional physician consults may be warranted.
12. You should be aware that there is no definitive cure for CF and other prenatal work-up maybe indicated if you and your partner are positive for the mutation(s). You should discuss this further with your health care provider.
13. No other test will be performed and/or reported on your sample unless authorized by your doctor. Any unused sample will be destroyed within 2-4 weeks of completion of this test.
14. The laboratory will disclose the test results only to your doctor or medical team, unless otherwise authorized by you or required by law.

I have read, or had read to me, the information about Cystic Fibrosis and I understand it. Before signing this form, I have had the opportunity to discuss the testing, its purpose and limitations, with my doctor or other member of the medical team. I was offered genetic counseling. I have all the information I need, and all of my questions have been answered. I have decided that:

____I do **not** want the CF carrier testing. ____I want the CF carrier testing.

_____ _____
Patient Signature Date

_____ _____ _____
Print Name of Guardian Guardian Signature Date

_____ _____ _____
Print Name of Physician/Genetic Counselor Signature Date